



Muscular Dystrophy New Zealand

November 2016

MYOTONIC DYSTROPHY – INFORMATION FACT SHEET

What is Myotonic Dystrophy?

Myotonic Dystrophy (DM) is an inherited disorder. It is abbreviated as DM as the Latin name for this condition is 'Dystrophia Myotonica'. In DM, a defective gene causes progressive muscle weakness accompanied by myotonia, which is the delayed relaxation of muscles after contraction. It is a multi-systemic disorder, meaning that its effects are not limited to the voluntary muscle system, but can also affect tissues and organs throughout the body. Primarily the muscles of the face, neck, hands, forearms and feet are affected, although DM can have a wide range of different effects on different individuals.

DM can be classified into several different types based upon the age of onset and the clinical symptoms experienced. Three main forms of DM have been identified so far:

- DM1, also known as Steinert's Disease
- DM2, also known as Proximal Myotonic Myopathy (PROMM)
- Congenital Myotonic Dystrophy (CMyD)

DM affects both males and females. Age of onset is variable from birth through to old age. It is a rare condition and DM1 and DM2 affects 1 in 8,000 individuals. CMyD is much rarer and affects 1 in 100,000.

The disorder has an effect called "anticipation" This means that DM can get progressively worse with each generation. In DM this effect is influenced by which parent, the mother or the father, passed on the condition.

What are the Features of Myotonic Dystrophy?

DM1

Myotonia will often be experienced as a stiffness or heaviness in the muscles with delayed relaxation, particularly in cold temperatures. This can occur in any of the muscles of the body. If myotonia significantly affects individuals, it can be treated with medication.

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Individuals typically present with muscle weakness and wasting. This is often apparent in the facial muscles of individuals with DM1, and can include a drooping of the eyelids (ptosis), an inability to fully close the eyes and to hold the jaw closed.

Severe ptosis can be aided with special glasses with 'eyelid crutches', which can be made by a skilled optician.

Surgery can also be performed, however weakness often returns. Neck muscles may also weaken, resulting in difficulty with lifting the head or sitting up quickly.

Weakness also often occurs in the distal muscles of the hands, lower legs and feet. They may experience difficulty in tasks requiring fine hand movements, such as writing. Individuals can experience gait disturbance (manner in which they walk) and may be prone to tripping due to foot drop, so have an exaggerated walk due to inability to control the foot at the ankle. Wrist supports, ankle-foot orthoses, canes and walkers can assist individuals where necessary, and can be discussed with a physiotherapist. Wheelchairs are rarely required, but may be beneficial for travelling long distances.

Respiratory (breathing) muscles can become weak, affecting the lungs and depriving the body of oxygen, which can contribute to individuals with DM1 feeling sleepy all the time. Regular sleep patterns can be helped with medication. To aid with breathing, a portable ventilator is commonly used.

Individuals may also experience difficulty swallowing (dysphagia) and can lead to choking. Vomiting can be very dangerous and sometimes fatal. A head-down position is crucial to prevent inhaling the vomit. A specialist can help people learn to swallow more safely and, if needed, how to change the consistencies of foods and liquids so they can be swallowed more easily.

Abnormalities of the conduction system in the heart are common and are a major cause of early mortality. The electrical signal that keeps the heart beating can get blocked and cause death. The most common signs are fainting or dizzy spells and these should not be ignored. Heart problems can be checked with an electrocardiogram (ECG) which measures the beating patterns of the heart. Severe problems can be corrected surgically with the insertion of a pacemaker.



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In DM, many of the body's internal involuntary muscles can weaken and have myotonia, including the digestive tract, the uterus and blood vessels. Spasms and weakness of the upper digestive tract can impair swallowing and it may feel like food is getting stuck. Occasionally food may be inhaled into the lungs. Care in swallowing, with advice from a physician or specialist may be necessary. Problems with the lower digestive tract may result in cramps, constipation or diarrhoea. Physicians can advise on management. Some drugs can help move things along the digestive tract if necessary.

Individuals with DM1 are at greater risk of developing gallstones due to weakening of the gallbladder – a sac under the liver which squeezes bile into the intestines. Symptoms are difficulty digesting fatty foods and pain in the upper right part of the abdomen and surgery is a possibility to remove the gallstones.

Women with DM1 may experience difficulties during pregnancy due to weakness and myotonia of the uterus. These may involve excessive bleeding or ineffective labour. Caesarean section may be advised.

Blood pressure tends to be low in individuals with DM1, however this does not usually pose a problem.

Some individuals may seem slow, dull, uncaring or depressed. As with most aspects of DM, there is a wide range of severity of the mental and emotional symptoms of the disorder. In adults, severe mental impairment is not very common, but an overall inability to "settle down to something," apply oneself to work or family life, concentrate or become engrossed in a task is often reported.

Cataracts – cloudy areas over the lens of the eye - are common in individuals with DM1. Individuals may notice haziness, blurriness or dimness that will worsen over time. This can be corrected with surgery.

Diabetes mellitus is a condition characterized by abnormally high blood and urinary sugar levels. Most individuals with DM1 suffer from a mild type referred to as 'insulin resistance'. Blood and/or urine tests may be carried out to test for this, and if present, individuals will often be advised to change diet and exercise habits or to take medication.



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Additional symptoms experienced in males may include premature balding, which can occur as early as 15 years of age, and testicular atrophy (loss of testicular size).

DM1 may be only mildly experienced and this form does not tend to impact significantly on an individual's life. They may only experience mild myotonia, cataracts, or diabetes mellitus. They can lead fully active lives and a normal or minimally shortened lifespan. Due to the mild nature of the symptoms, DM often goes undiagnosed.

DM2 – Proximal Myotonic Myopathy

DM2 is clinically similar to DM1 and tends to have a differing pattern of muscle weakness. There is greater weakness in proximal limb muscles, such as the upper leg, and less so in the face. This can result in difficulty in climbing stairs or rising from the floor. Myotonia is often mild or not present at all. Individuals with DM2 may have minor cognitive difficulties. Cataract, heart problems and diabetes also occur as in DM1, while other complications are rare.

Congenital Myotonic Dystrophy

Congenital myotonic dystrophy is a form of DM that is present at birth, and only occurs in individuals with mothers with DM1. In many instances, the mother's condition is so mild that having a baby with congenital myotonic dystrophy is the first indication that the mother has DM. Congenital myotonic dystrophy is significantly more severe than other DM1 or 2 and affects boys and girls equally.

Note: Mothers with DM can also pass on the DM1 or DM2 form. A child can also inherit myotonic dystrophy from the father, but it's almost always the DM1 or DM2 form.

Babies with Congenital myotonic dystrophy have severe muscle weakness and hypotonia (loss of muscle tone). This includes weakness of the face and children characteristically have a 'tented' upper-lip. Babies are also often born with clubfeet – a curvature of the feet and lower legs. Surgical correction is necessary to enable the child to walk. The muscles that control breathing and swallowing are also involved. These problems can be life-threatening and need early intervention and intensive care to improve the chance of survival. This may include the use of artificial ventilation and a feeding tube inserted into the stomach.



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Muscles used for speech are often affected and hearing can also be impaired. Speech therapists may be beneficial. Weakness of the eye muscles can result in strabismus, a condition where the eyes do not work together. This can be corrected with surgery.

Myotonia is not present in babies with congenital myotonic dystrophy, however if the infant survives, he or she may develop myotonia later in life. Motor function gradually improves in surviving children and they are usually able to walk. However they will develop the same clinical signs of DM1 later in life.

Congenital myotonic dystrophy may also result in severe learning problems and sometimes an individual may be mentally retarded. Special education is often necessary because of these disabilities.

Additional Considerations for Myotonic Dystrophy

An unusually high rate of complications and even deaths associated with general anaesthesia (given during any surgery) has been reported in people with DM. Surgery can usually be safely undertaken with careful monitoring of cardiac and respiratory functions before, during and after the surgery. Be sure to tell the entire medical team, especially those responsible for the anaesthesia, that you or your family member has DM.

What Causes Myotonic Dystrophy?

All types of DM are autosomal dominant disorders. For further information on genetics and how disorders are inherited, please refer to the Muscular Dystrophy Association Genetics Factsheet.

Humans have 46 chromosomes made up of genes. Each chromosome, which is a tightly coiled chain of DNA (deoxyribose nucleic acid) contains millions of chemicals called bases. The four bases are adenine, thymine, cytosine and guanine (A, T, C and G), which pair together in sets of three to form coded messages. These messages are instructions for producing proteins that make the body function. Many disorders are a result of a mutation to these bases.

In all forms of DM, there is an abnormal expansion of different sequences of bases. In DM1 and Congenital Myotonic Dystrophy it is a result of an expanded repeat of the CTG sequence in the DMPK (dystrophica myotonica protein kinase) gene on chromosome 19. DM2 is a result of an expanded repeat of CCTG in the ZNF9 (zinc finger protein 9) on chromosome 3. Generally a greater expansion is related to earlier onset and faster progression of the disorder.

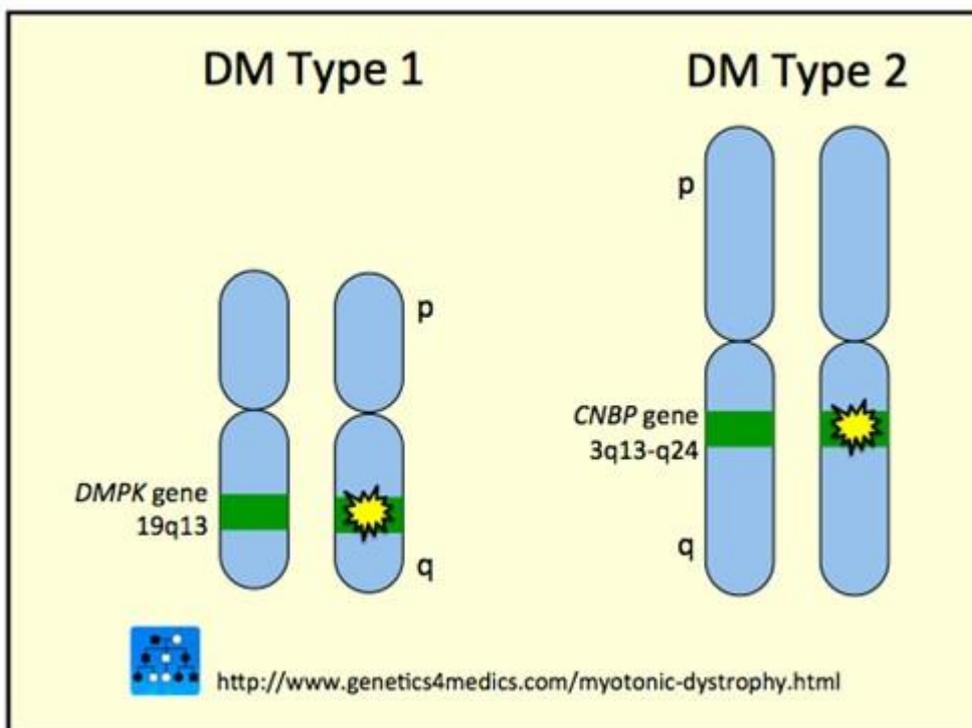
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How these mutations directly affect all the different systems of individuals is currently unknown, however researchers have hypothesized that the problem lies in RNA, which is a copied version of DNA for processing genes into proteins. When the expanded sequence in the DNA is copied, there is an over-accumulation of RNA which traps the information inside the center of the cell. This blocks several other types of RNA and disrupts the protein-manufacturing process for the genes which control several bodily processes.





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Diagnosis of Myotonic Dystrophy

Diagnosis usually commences after the identification of key early symptoms of DM:

- 'Grip Test' - affected individuals will not be able to open and close their hand rapidly and will have a characteristic grip
- Blood Testing – elevated levels of creatine phosphokinase (CPK) are indicative of muscle problems
- Electromyography (EMG) – observes the electrical activity of muscles and its consistency with activity typical of DM individuals
- Muscle Biopsy – looks at individuals muscle cells for characteristic features of DM
- DNA Testing – can identify the presence of the abnormal gene in the individual with DM

Soon after a diagnosis of DM in the family, it is essential that genetic counselling is arranged, for one or both of two issues. The first is the probability of Mum or Dad having the disorder, and the second is whether testing for DM in pregnancy can be offered and with what degree of accuracy.

Genetic counselling provides information about possible diagnostic tests, including prenatal testing. Genetic services in NZ are available and a referral can be made by the MDA.

Management of Myotonic Dystrophy

As there is currently no cure for DM, treatment focuses on the prevention and management of symptoms which, as explained above, differ from one individual to another, dependent on how severely he or she is affected.

Research into Myotonic Dystrophy

Research is continuously being carried out and as more discoveries are made about the underlying causes of DM, it will be possible to develop treatments aimed more directly at these causes, rather than simply towards the management of its symptoms.

Support for People with Myotonic Dystrophy

Support is available from the MDA who can offer specialist information, support, advocacy and referrals to other providers. There is also a nationwide Support Network for those interested in meeting with others.

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There is no reason why individuals with DM should be disadvantaged in terms of receiving full education. For more information, request the Education Pack available from the MDA.

Disability should not hinder employment possibilities. Any individual has the right to equal pay and equal rights for employment. For more information contact the Employment Relations infoline on [0800 800 863](tel:0800800863) or visit www.ers.dol.govt.nz.

The government promotes equal employment opportunities in private sector and can be contacted on [\(09\) 525 3023](tel:095253023) or visit www.eeotrust.org.nz

Workbridge provides a professional employment service for people with all types of disabilities and administers support funding on behalf of Work and Income. Contact on [0508 858 858](tel:0508858858) or visit www.workbridge.co.nz

More Information

Muscular Dystrophy Association can be contacted for further information, assistance, advice, support and referrals, on [0800 800 337](tel:0800800337) or by e-mail at info@mda.org.nz. The Muscular Dystrophy Association Website also contains information on services available within NZ, a quarterly magazine, contacts, membership details, news and links to other sites - www.mda.org.nz

Further Resources

www.nzord.org.nz – the New Zealand Organisation for Rare Disorders website provides information on a number of rare disorders, a directory of support groups, practical advice, health and disability resources, research information, news and issues.

www.mdaua.org – the MDA USA website has an extensive site with plenty of further information on any muscular dystrophy conditions as well as research news.

www.myotonicdystrophy.org – International Myotonic Dystrophy Organization has further information on Myotonic Dystrophy and its management, recent research, support groups and forums.

Information in this fact sheet was primarily sourced from: Muscular Dystrophy Association USA (2005) Facts About Myotonic Muscular Dystrophy. www.mdaua.org

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